Name ____

Investigation 2: Hardy-Weinberg
Population Genetics and Evolution

Objectives

- Estimate the frequency of alleles in a population using the Hardy-Weinberg equations
- Demonstrate that allele frequencies can change in a population over time

Background

In the early 1900s, many biologists attempted to explain evolution in terms of the emerging science of genetics. Because the F_2 of a monohybrid cross show a 3:1 ratio of dominant to recessive phenotypes, many assumed that populations would evolve toward similar ratios of phenotypes. Two mathematicians, Godfrey Hardy and Wilhelm Weinberg, realized that the frequency of alleles in a population was independent of the alleles' inheritance pattern from individual parents to offspring. They postulated an ideal breeding population with the following properties:

1. Population size is infinite, or very large.
2. Mating is random. Mating pairs show no preference for one phenotype over another.
3. There is no mutation.
4. There is no exchange with other populations. No immigration or emigration occurs.
5. There is no selection for one phenotype over another. All phenotypes have an equal chance of reproducing.

If these conditions are met, the populations, allele and genotype frequencies will remain statistically constant over time, a condition referred to as Hardy-Weinberg equilibrium. If we determine the frequency of a pair of alleles in a population, we can sample that population over several generations to determine if the frequency changes. If it does, we know that the population is evolving with respect to that pair of alleles.

Hardy and Weinberg developed mathematical formulas for estimating the frequencies of alleles in populations.

$$p + q = 1$$
$$p^2 + 2pq + q^2 = 1$$

p = the frequency of the dominant allele (represented here by A)
q = the frequency of the recessive allele (represented here by a)

p^2 = frequency of AA (homozygous dominant)
$2pq$ = frequency of Aa (heterozygous)
q^2 = frequency of aa (homozygous recessive)

Name ____

Procedures

Activity A: A Test of Hardy-Weinberg Equilibrium

In this activity, the class will simulate a breeding population of diploid organisms. Each individual will have four cards, which represent gametes produced by meiosis. (Remember that gametes produced by meiosis are haploid.) The letter on the card represents an allele that is inherited with the gamete. You will contribute one gamete to each of your offspring. Everyone in the class will begin with the same four cards, two A and two a.

1. Determine the initial frequencies of alleles A and a and record them in the results section. (Be sure to show your work!)
2. Shuffle your cards face-down. Pair with another student. Draw the top card from your hand and match it with your partner's card. These two cards represent the genotype of your first offspring. One of you should record this as the Generation 1 genotype in Table 1.
3. Retrieve your card and reshuffle your hand. Repeat Step 2 with the same partner to produce a second offspring. The individual who did not record a Generation 1 Genotype in Step 1 should record this result in Table 1 for Generation 1.
4. Now, assume the genotype of your Generation 1. For example:
   - If you recorded the genotype aa, you should have four cards, all marked a
   - If you recorded the genotype AA, you should have four cards, all marked A
   - If you recorded the genotype Aa, keep your original hand of cards (two marked A and two marked a).

   Discard and obtain cards as necessary.
5. Randomly pair with a different student and repeat this process for five generations.
6. Record the class totals for each genotype in Table 1.
7. Calculate the Generation 5 class frequencies of each allele and record them in the results section. (Be sure to show your work!) Round to the nearest tenth.

Name ____

Activity B: Directional Selection

A genotype can affect the reproductive success of the organism that carries it. For example, a recessive allele results in the production of an abnormal hemoglobin molecule, hemoglobin S. People who are homozygous for this recessive allele have the disease sickle-cell anemia. Historically, people with sickle-cell anemia have rarely survived childhood and thus have rarely reproduced. What happens to such an allele in a population over time? You will explore this question in the following simulation. In this activity, you will assume that none of the homozygous recessive individuals (aa) reproduce and that all AA and Aa individuals do reproduce.

1. As before, being with four cards, two A and two a.
2. Follow the steps take in Activity A once again, but with the following exception: do not record any aa offspring.
    - When the aa combination results, retrieve your cards, reshuffle your hand, and try again until you get the AA or the Aa combination.
3. Repeat this process through five generations to complete Table 2.
4. Once again, record the class totals for each genotype.
5. Calculate the Generation 5 class frequencies of each allele and record them in the results section. (Be sure to show your work!) Round to the nearest tenth.

Activity C: Heterozygote Advantage

Studies indicate that individuals who are heterozygous for the hemoglobin S allele, Aa, may have an increased resistance to malaria. What happens to the recessive allele in a population when there is selection against both homozygotes (AA and Aa)? The following simulation explores this question. To better reveal the results of selection, you will simulate more generations in this activity.

1. Once again, begin with four cards, two A and two a.
2. Follow the procedure in Activity B, with this addition condition: if the offspring is AA, flip a coin.
    - If the result is heads, the offspring survives
    - If the result is tails, the individual does not survive
        - In this case retrieve your card, reshuffle your hand, and try again

    Remember, as in Activity B, the genotype aa never survives!
3. Repeat this process until you produce two surviving offspring each generation for TEN generations. Record your results in Table 3.
4. Once again, record the class totals for each genotype.
5. Calculate the Generation 10 class frequencies of each allele and record them in the results section. (Be sure to show your work!) Round to the nearest tenth.

Name ____

Activity D: Genetic Drift

It is possible to use our simulation to look at the phenomenon of genetic drift in detail.

1. You will divided into three smaller, isolated populations. Individuals from one population will not interact with individuals from other populations.
2. You will follow the procedure used in Activity A, starting with 2 A cards and 2 a cards. (Once again, all offspring survive and reproduce.) Go through five generations and record your data in Table 4.
3. Instead of recording class totals for each genotype, we will be recording the totals for each subpopulation in Table 5.
4. Calculate and compare the Generation 5 frequencies of each allele for each of the 3 subpopulations and record them in Table 6. (Still be sure to show your work!) Round to the nearest tenth.

Results

Activity A: A Test of Hardy-Weinberg Equilibrium

Initial Allele Frequencies: The allele frequencies, p and q, should be calculated for the class population BEFORE five generations of simulated random mating.

Number of A alleles present to start

Number of offspring with genotype AA _________ x 2 = ________ A alleles
Number of offspring with genotype Aa _________ x 1 = ________ A alleles
Total = ________ A alleles

$$p = \frac{\text{TOTAL number of A alleles}}{\text{TOTAL number of alleles in the population (number of students x 2)}} = \boxed{}$$

Number of a alleles present to start

Number of offspring with genotype aa _________ x 2 = ________ a alleles
Number of offspring with genotype Aa _________ x 1 = ________ a alleles
Total = ________ a alleles

$$q = \frac{\text{TOTAL number of a alleles}}{\text{TOTAL number of alleles in the population (number of students x 2)}} = \boxed{}$$

Name ____

Table 1: Hardy-Weinberg Equilibrium

Initial Genotype **Aa**	My Genotype	Class Totals		
		AA	Aa	aa
Generation 1				
Generation 2				
Generation 3				
Generation 4				
Generation 5				

Final Allele Frequencies: The allele frequencies, p and q, should be calculated for the class population AFTER five generations of simulated random mating.

Number of A alleles present at the 5th generation

Number of offspring with genotype AA _________ x 2 = ________ A alleles
Number of offspring with genotype Aa _________ x 1 = ________ A alleles
Total = ________ A alleles

$$p = \frac{\text{TOTAL number of A alleles}}{\substack{\text{TOTAL number of alleles in the population} \\ \text{(number of students x 2)}}} = \boxed{}$$

Number of a alleles present at the 5th generation

Number of offspring with genotype aa _________ x 2 = ________ a alleles
Number of offspring with genotype Aa _________ x 1 = ________ a alleles
Total = ________ a alleles

$$q = \frac{\text{TOTAL number of a alleles}}{\substack{\text{TOTAL number of alleles in the population} \\ \text{(number of students x 2)}}} = \boxed{}$$

Name ____

Activity B: Directional Selection

Table 2: Selection

Initial Genotype **Aa**	My Genotype	Class Totals		
		AA	Aa	aa
Generation 1				
Generation 2				
Generation 3				
Generation 4				
Generation 5				

Final Allele Frequencies: The allele frequencies, p and q, should be calculated for the class population AFTER five generations of simulated random mating.

Number of A alleles present at the 5th generation

Number of offspring with genotype AA __________ x 2 = _________ A alleles
Number of offspring with genotype Aa __________ x 1 = _________ A alleles
Total = _________ A alleles

$$p = \frac{\text{TOTAL number of A alleles}}{\text{TOTAL number of alleles in the population (number of students x 2)}} = \square$$

Number of a alleles present at the 5th generation

Number of offspring with genotype aa __________ x 2 = _________ a alleles
Number of offspring with genotype Aa __________ x 1 = _________ a alleles
Total = _________ a alleles

$$q = \frac{\text{TOTAL number of a alleles}}{\text{TOTAL number of alleles in the population (number of students x 2)}} = \square$$

Name ____

Activity C: Heterozygote Advantage

Table 3: Heterozygote Advantage

Initial Genotype **Aa**	My Genotype	Class Totals		
		AA	Aa	aa
Generation 1				
Generation 2				
Generation 3				
Generation 4				
Generation 5				
Generation 6				
Generation 7				
Generation 8				
Generation 9				
Generation 10				

Final Allele Frequencies: The allele frequencies, p and q, should be calculated for the class population AFTER ten generations of simulated random mating.

Number of A alleles present at the 10th generation

Number of offspring with genotype AA __________ x 2 = _________ A alleles
Number of offspring with genotype Aa __________ x 1 = _________ A alleles
Total = _________ A alleles

$$p = \frac{\text{TOTAL number of A alleles}}{\text{TOTAL number of alleles in the population (number of students x 2)}} = \square$$

Number of a alleles present at the 10th generation

Number of offspring with genotype aa __________ x 2 = _________ a alleles
Number of offspring with genotype Aa __________ x 1 = _________ a alleles
Total = _________ a alleles

$$q = \frac{\text{TOTAL number of a alleles}}{\text{TOTAL number of alleles in the population (number of students x 2)}} = \square$$

Name ____

Activity D: Genetic Drift

Table 4: Genetic Drift (Individual Data)

Initial Genotype **Aa**	My Genotype
Generation 1	
Generation 2	
Generation 3	
Generation 4	
Generation 5	

Table 5: Genetic Drift (Class Data divided into 3 isolated subpopulations)

Initial Genotype **Aa**	Subpopulation 1 Totals		
	AA	Aa	aa
Generation 1			
Generation 2			
Generation 3			
Generation 4			
Generation 5			

Initial Genotype **Aa**	Subpopulation 2 Totals		
	AA	Aa	aa
Generation 1			
Generation 2			
Generation 3			
Generation 4			
Generation 5			

Initial Genotype **Aa**	Subpopulation 3 Totals		
	AA	Aa	aa
Generation 1			
Generation 2			
Generation 3			
Generation 4			
Generation 5			

Table 6: Genetic Drift Class Frequencies

	Allele Frequencies		
	Subpopulation 1	Subpopulation 2	Subpopulation 3
p			
q			

Name ____

Show Work for Allele Frequencies Here:

Analysis

Activity A: A Test of Hardy-Weinberg Equilibrium

1. Are the generation 5 values for p and q drastically different from their initial values? Is this consistent with the alleles being at equilibrium? If not, why not?

2. Review the five conditions that must be met for allele frequencies to remain constant. Which, if any, of these conditions might not have been met in this simulation?

Name ____

Activity B: Directional Selection

3. Are the generation 5 values for p and q different from their initial values? Explain why the values did or did not change.

4. What happened to the Generation 5 values for p and q you just calculated in Activity B compared with those you calculated in Activity A?

5. This simulation involved very high selection against the recessive allele. Did selection eliminate the allele from the population? If not, why did the allele persist?

Activity C: Heterozygote Advantage

6. What happened to the final frequencies for p and q you just calculated in Activity C compared with those you calculated in Activity B? Explain why they are different.

Name ____

7. Consider two isolated populations of people who have occupied their homelands for untold generations. One population occupies a dry, windswept plain. The other populations inhabits a lowland, tropical area with many streams and swamps. In which population would you expect to find the higher frequency of the allele for hemoglobin S? Explain your reasoning. (Hint: Mosquitoes, which are vectors for and thus spread malaria, need stagnant water to complete their life cycle.)

8. Is the mutation for hemoglobin S harmful or beneficial? Support your answer.

Activity D: Genetic Drift

9. How do the final genotypic frequencies of EACH population compare? Explain why.

10. What do the class results indicate about the importance of population size as an evolutionary force?

Name ____

Problems

1. In *Drosophilia* the allele for normal-length wings is dominant over the allele for vestigial wings (vestigial wings are stubby little curls that cannot be used for flight). In a population of 1,000 individuals, 360 show the recessive phenotype. How many individuals would you expect to be homozygous dominant and heterozygous for this trait?

2. The allele for the hair pattern called "widow's peak" is dominant over the allele for no "widow's peak." In a population of 1,000 individuals, 510 show the dominant phenotype. How many individuals would you expect of each of the possible three genotypes for this trait?

Name ____

3. The allele for unattached earlobes is dominant over the allele for attached earlobes. In a population of 500 individuals, 25% show the recessive phenotype. How many individuals would you expect to be homozygous dominant and heterozygous for this trait?

4. In the United States, about 16% of the population is Rh negative. The allele for Rh negative is recessive to the allele for Rh positive. If the student population of a high school in the U.S. is 2,000, how many students would you expect for each of the three possible genotypes?

5. In a certain population, the dominant **phenotype** of a certain trait occurs 91% of the time. What is the frequency of the dominant allele?